BIOLOGY

EST, SAT, ACT PREP

DR MAHMOUD FAHMY

MOBILE: 01090467169

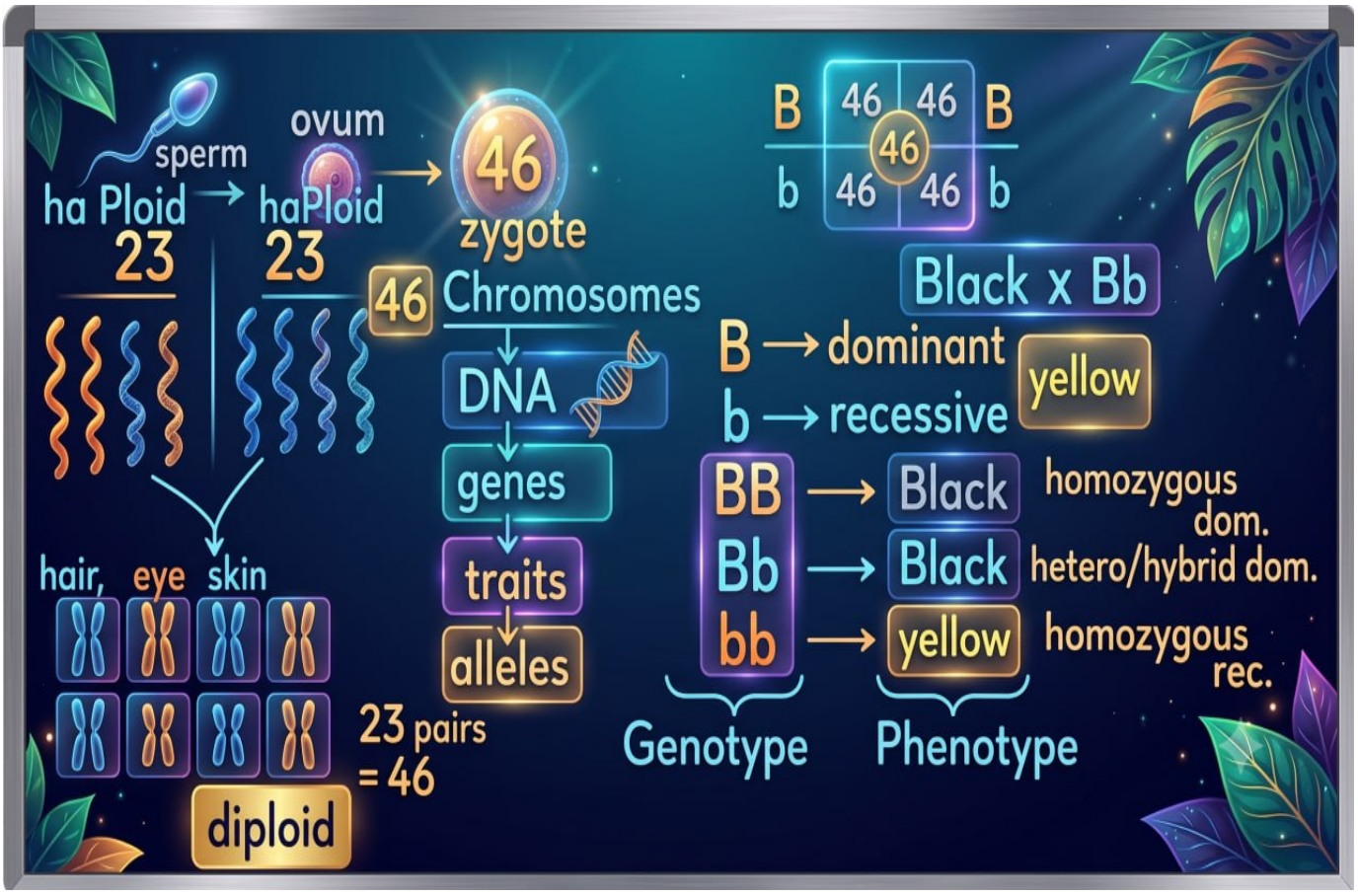
DR MAHMOUD FAHMY 01090467169

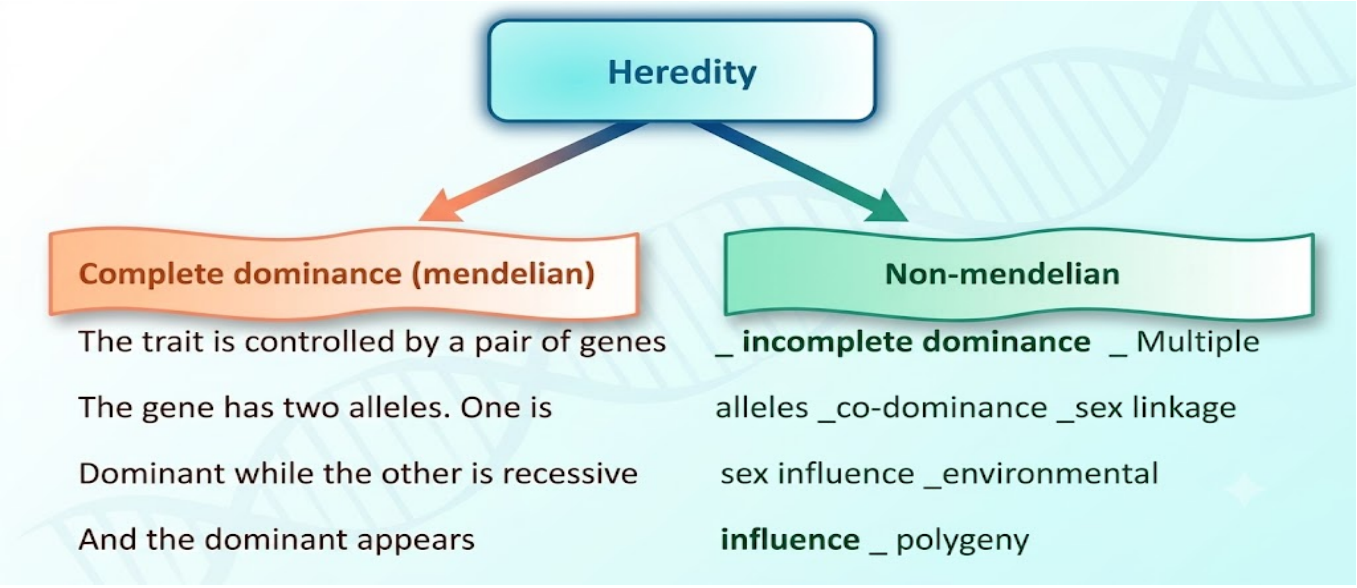
Chapter 3: Classical Genetics

Important Genetics Terminology

- Diploid (2N): Cells in which chromosomes are found in pairs. Example: Body cells.
- Haploid cells (N): Cells in which chromosomes are single. Example: Gametes (sperm and ova) and bacteria.
- Chromosome: DNA + Histone Protein.
- Homologous chromosomes: Paired chromosomes.
- Gene (hereditary factor): Part of DNA that carries a code for a certain characteristic.
- Allele: Alternative forms of the same gene (e.g., brown and blue eye colors).
- Homozygous: Pure (similar factors).
- Heterozygous: Impure or hybrid (different factors).
- Dominant: Masking allele.
- Recessive: Masked allele.
- Phenotype: External appearance.
- Genotype: Genetic make-up.

Note: Recessive individuals are always pure. In the case of recessive traits, the phenotype indicates the genotype, while in the case of dominant traits, the phenotype doesn't indicate the genotype.




Heredity
Complete dominance (mendelian)
Non-mendelian
The trait is controlled by a pair of genes
The gene has two alleles. One is
Dominant while the other is recessive
And the dominant appears
_ incomplete dominance _ Multiple
alleles _co-dominance _sex linkage
sex influence _environmental
influence _ polygeny

Mendel's first law: law of segregation (monohybrid cross)

The law states that when two pure organisms for two opposing traits are crossed, the offspring will be hybrid but will exhibit only the dominant trait while the recessive trait remains hidden.

P TT (Long) x tt (Short)
G T T t t
F1 Tt, Tt, Tt, Tt
100% Long
P Tt x tt
G T t t t
F1 Tt, Tt, tt, tt
50% Long, 50% Short
Allele Key
T = Long (Green text)
t = Short
F2 Monohybrid Cross Analysis
P2 Tt x Tt
G2 T t T t
F2 TT Tt Tt tt
Long Long Long Short
0 = 0%
¼ = 25%
½ = 50%
¾ = 75%
1 = 100%
Genotype Ratio 1 (TT) : 2 (Tt) : 1 (tt)
Phenotype Ratio 3 (Long) : : 1 (Short)
1 homo dom x homo rec = 100% hetero
BB x bb = 100% Bb
2 hetero x hetero = 3:1 (Phenotype)
Bb x Bb = 3 Black : 1 Yellow
3 test cross hetero x homo rec = 1:1 (Phenotype)
Bb x bb = 50% Black : 50% Yellow

Advantages of Mendel's Work:

- He studied the transmission of traits for more than one generation.
- He introduced quantitative study in biology.

Backcross or testcross

• A method to distinguish the genotype of the dominant individual whether it is homo or hetero
• It's made by crossing it with recessive (aa or tt)

It's made by crossing it with recessive

The Result of offspring	
100%	So the individual was homozygous
50% : 50% (1:1)	So the individual was heterozygous

P: TT x t t

G: (T) (T) (t) (t)

F_1: Tt Tt Tt Tt 100%

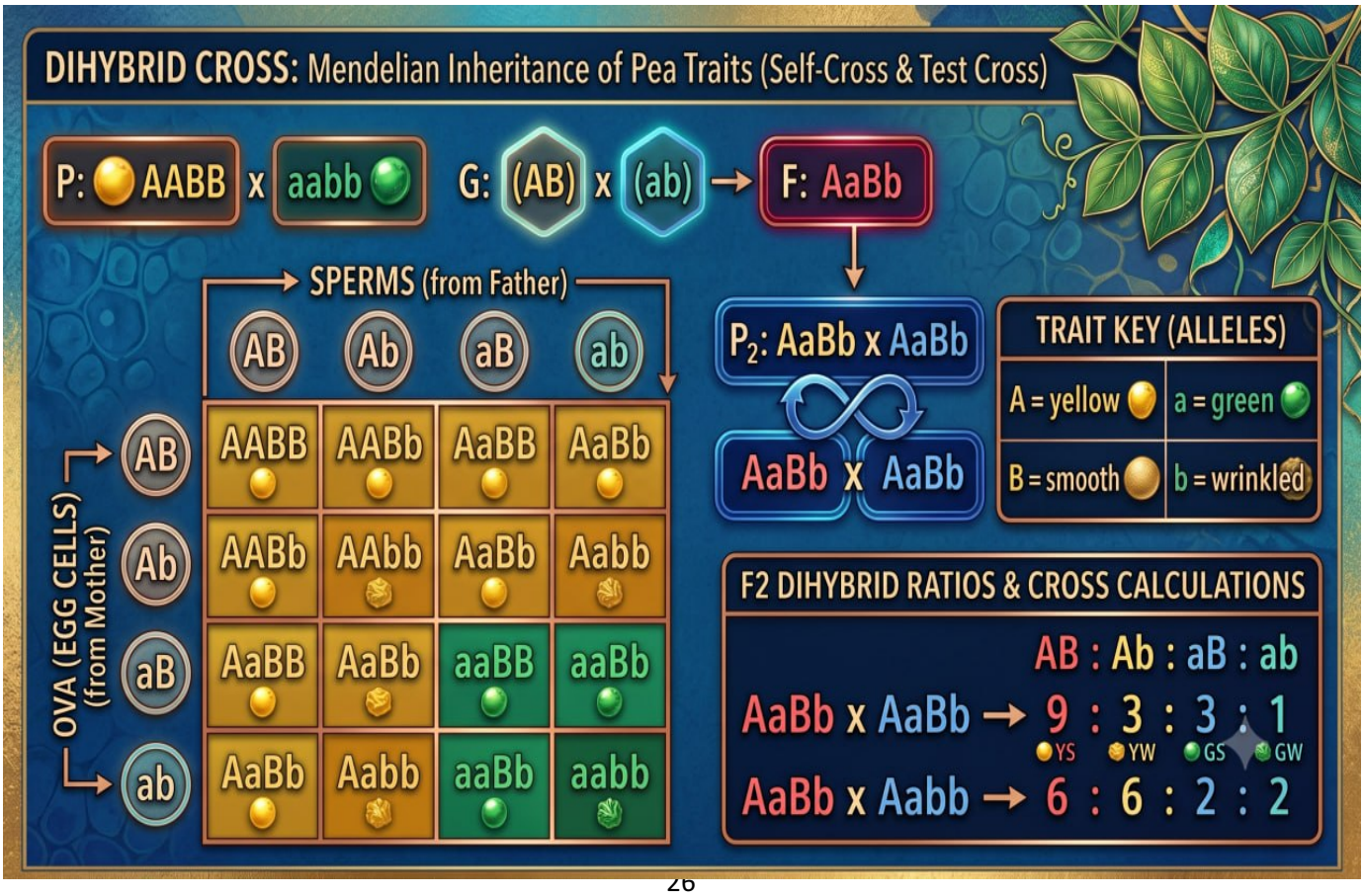
P: Tt x tt

G: (T) (t) (t) (t)

F_1: Tt Tt tt tt

1 : 1

Mendel's second law: law of independent assortment (dihybrid inheritance)

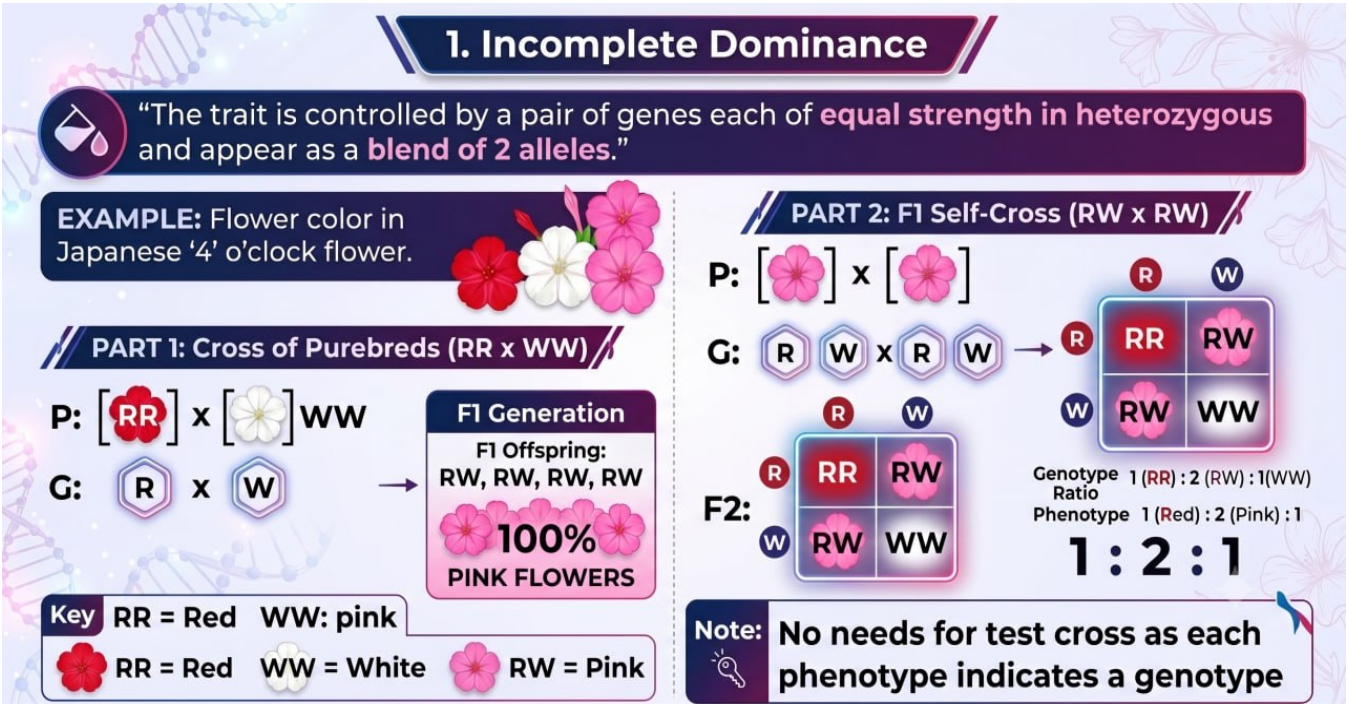

DIHYBRID CROSS: Mendelian Inheritance of Pea Traits (Self-Cross & Test Cross)
P: AABB x aabb
G: (AB) x (ab)
F: AaBb
SPERMS (from Father)
AB
Ab
aB
ab
OVA (EGG CELLS) (from Mother)
AABB AABb AaBB AaBb
AABb AAbb AaBb Aabb
AaBB AaBb aaBB aaBb
AaBb Aabb aaBb aabb
P2: AaBb x AaBb
AaBb x AaBb
TRAIT KEY (ALLELES)
A = yellow
a = green
B = smooth
b = wrinkled
F2 DIHYBRID RATIOS & CROSS CALCULATIONS
AB : Ab : aB : ab
AaBb x AaBb → 9 : 3 : 3 : 1
YS YW GS GW
AaBb x Aabb → 6 : 6 : 2 : 2

• **AABB → one gametes**
• **AaBB → two gametes**
• **AaBb → four gametes**
• **Mendel didn't explain crossing over or phylogeny or chromosomal maps**
• **True-breeding → all members have the same phenotype**
• **Aa × aa → 2 phenotypes**
• **AaBb × AaBb → 4 phenotypes**

Non-Mendelian

I. Incomplete dominance

• The trait is controlled by a pair of genes each of equal strength in heterozygous and appear as a blend of 2 alleles
• 3 phenotypes (as 3 colors)
• Ratio → **1 : 2 : 1**


1. Incomplete Dominance
"The trait is controlled by a pair of genes each of equal strength in heterozygous and appear as a blend of 2 alleles."
EXAMPLE: Flower color in Japanese '4' o'clock flower.
PART 1: Cross of Purebreds (RR x WW)
P: RR x WW
G: R x W
F1 Generation
F1 Offspring: RW, RW, RW, RW
100% PINK FLOWERS
Key RR = Red WW: pink
RR = Red WW = White RW = Pink
PART 2: F1 Self-Cross (RW x RW)
P: x
G: R W x R W
F2: RR RW RW WW
Genotype Ratio 1 (RR) : 2 (RW) : 1(WW)
Phenotype 1 (Red) : 2 (Pink) : 1
1 : 2 : 1
Note: No needs for test cross as each phenotype indicates a genotype

II. Co-dominance

• The trait is controlled by a pair of genes, each gene has 2 alleles, equal in strength in the heterozygous state
• Each gene expresses itself equally and independently

Example:
Blood group AB (A + B both appear).

- Spotted animals Speckled chicken (black and white feathers together).

DR. MAHMOUD FAHMY 01122666893

III. Polygenes (A case of incomplete dominance with many genes)

- The trait is controlled by many pairs of genes
- The degree of the trait depends on the number of each allele
- e.g. skin color & body height

IV. Multiple alleles

"The trait is controlled by a pair of genes; each gene has more than 2 alleles"

Ex: ABO blood type

Antigen	Antibody
Special proteins found on the surface of RBCs that determines the blood group	Antithetic to antigene found in blood plasma

Blood group	phenotype	genotype
A	A	AA(I^AI^A), AO (I^AI^O)
B	B	BB (I^BI^B), BO (I^BI^O)
AB	AB	AB (I^AI^B)
O	O	OO (I^OI^O)

Blood Group	Antigen	Antibody
A	A	Anti-b
B	B	Anti-a
AB	A , B	--
O	--	Anti-a, Anti-b

Blood transfusion:

Blood Group	A	B	AB	O
Donates to	A , AB	B , AB	AB	Universal
Receive from	A , O	B , O	Universal	O

DR. MAHMOUD FAHMY 01122666893

Denying Paternity

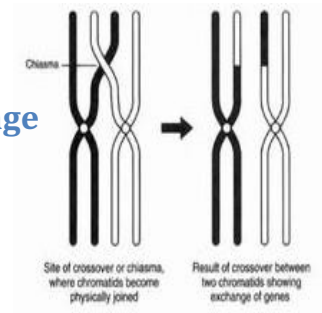
Case: The blood group of a baby is O , we want to know the baby belongs to which parent. if the two wives have blood group O but the two fathers are different, the 1st father has A blood group while the 2nd father has AB blood group

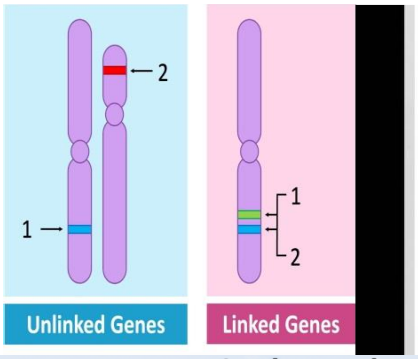
P: AA x OO	P: AO x OO	P: AB x OO
G: (A)(A) (O)(O)	G: (A)(O) (O)(O)	G: (A)(B) (O)(O)
F: AO AO AO AO	F: AO AO OO OO	F: AO AO BO BO
The baby may be O with 50%		No babies are OO

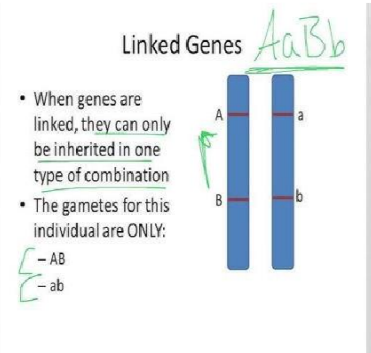
So the baby belongs to the first parents.

- *Father with AB can't get O & vice versa*
- *Hetero (AO) + hetero (BO) → all blood types*
- *Rh factor is a protein found in blood plasma*
- *Rh^+ means that it is present (dominant)*
- *Rh^- means that it is absent (recessive)*
- *Rh^+Rh^+ → positive , Rh^+Rh^- → positive , Rh^-Rh^- → Negative*
- *Rh^- cannot receive blood from Rh^+ as he will produce antibodies*
- *Incorrect blood transfusion → the blood agglutinates (O is the safest one)*

V. Linkage


Chiasma
Site of crossover or chiasma, where chromatids become physically joined
Result of crossover between two chromatids showing exchange of genes





Linked genes: are genes carried on the same chromosone

Ex: body color and wing shape of drosophila

G = grey g = black L = long wings l = short wings

P: GG LL x gg ll

G: (GL) (g l)

F1: GgLl 100% grey long winged

P: GgLl x GgLl

G: (GL) (gl) (GL) (gl)

F2: GGLL GgLl GgLl ggll

3 : 1

• ***AaBb → two gametes only (remember Mendel's second law)***
• ***If he gave you 2 traits then check the ratio:***
– ***If it is 9 : 3 : 3 : 1 → independent assortment***
– ***If it is 3 : 1 → then linkage***
• ***Linked genes can be separated during crossing over (exchange of segment of chromosomes)***
• ***DNA exchange segments → recombination***

VI. Sex linkage

Sex Determination in Humans

Female: 44 + XX - Meiosis⟶ 22 + X
Male: 44 + XY - Meiosis⟶ 22 + X or 22 + Y

Types of cells

Somatic cells	Reproductive cells
Body cells Contain diploid no. of chromosomes 44 + XX or 44 + XY	Sex cells (gametes) Contain haploid no. of chromosomes 22 + X or 22 + Y

Types of chromosomes

Autosomes (Somatic chromosomes)	Sex chromosomes
Carries traits that are similar in male and female	Carries sex determining genes

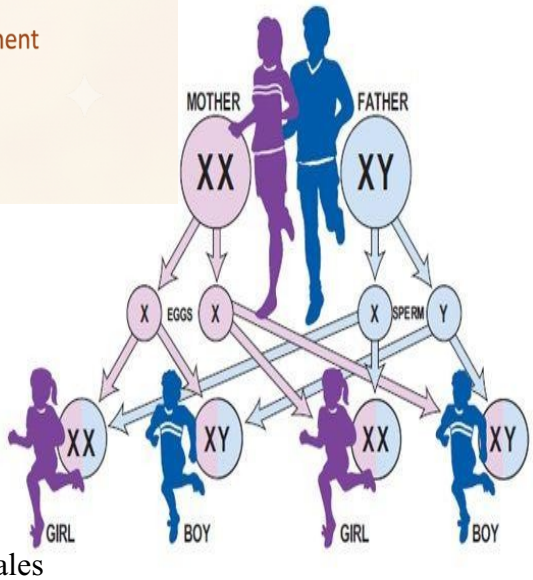
The difference between X and Y chromosomes:

- ❖ X chromosome is bigger so it carries more genes
- ❖ X chromosome has other genes responsible for growth and development

Sex linked genes = X linked gene

Ex: Color blindness and haemophilia

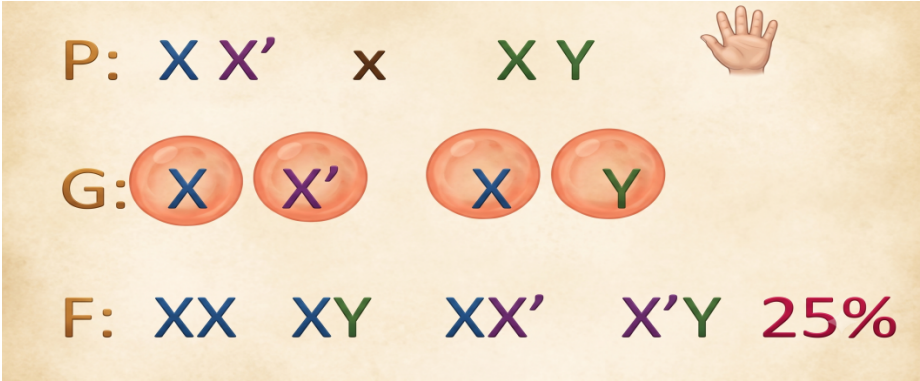
e.g. hemophilia & color blindness



• X = Normal (dominant) , X′ = infected (recessive)
• In Females:
– X X = normal – X X′ = carrier – X′ X′ = infected
• In Males:
– X Y = normal
– X′ Y = infected
• No male carriers for X-linked traits (has only one X)
• X-linked recessive traits are more common in males than in females
• Sons cannot inherit sex-linked traits from the fathers because the sons inherit Y chromosomes from their fathers

DR. MAHMOUD FAHMY 01122666893

Example: A woman with normal color vision, whose father was color-blind, married a man with normal color vision. What is the probability that they have a son who is color-blind?



VII. Sex influence

7. Sex-influenced Traits

Sex-influenced traits are autosomal traits that are expressed based on the influence of sex.

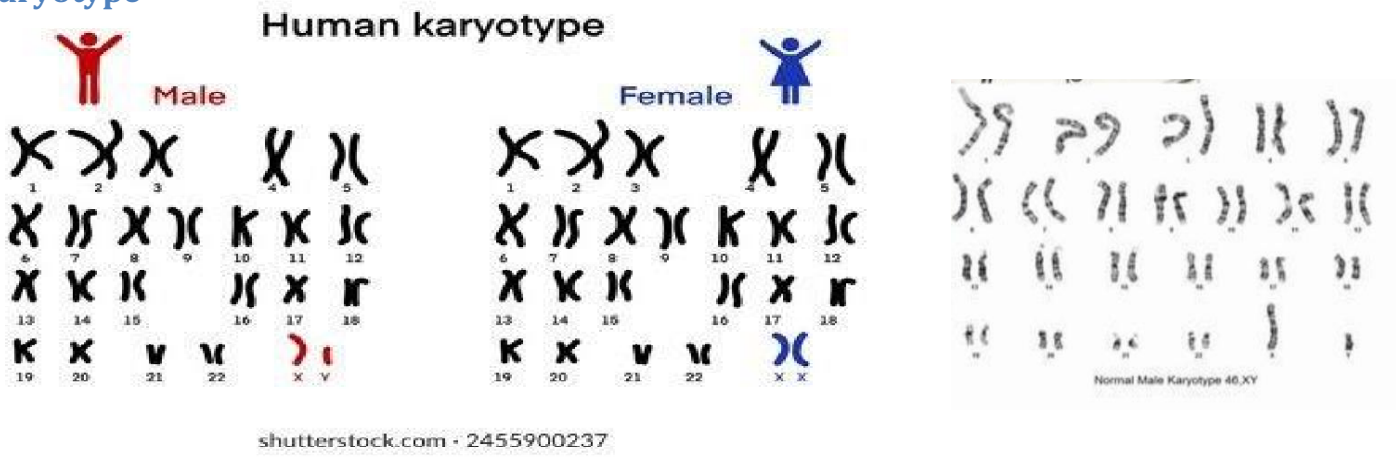
Example: Premature baldness
- Testosterone causes the baldness gene to be dominant in males.
- Males express baldness more frequently than females.

8. Environmental Influence

The environment can alter the expression of genes.

Examples:
- Fruit flies in hot environments develop long wings.
- White-furred Himalayan rabbits develop black fur in cold temperatures.

Karyotype

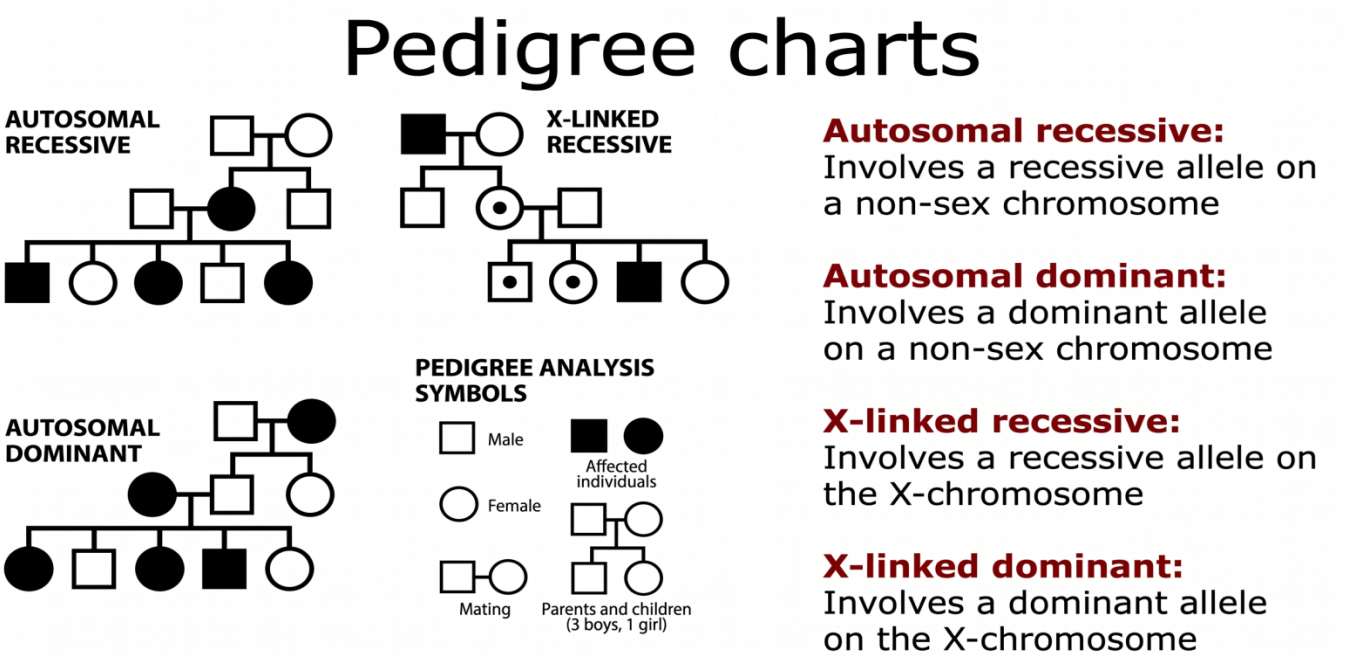


- Is a laboratory procedure that analyzes the size, shape and number of chromosomes
- Or "arrangement of chromosomes according to their size"
- Specialists photograph chromosomes during the metaphase of mitosis when they are fully condense

Pedigree

• Is a family tree that indicates the phenotype of one trait being studied for every member of a family

- Females are represented by circles and males by squares.
- The carrier state is sometimes represented by a half-shaded shape.
- A shape is completely shaded if the individual exhibits the trait.


Pedigree charts
AUTOSOMAL RECESSIVE
X-LINKED RECESSIVE
AUTOSOMAL DOMINANT
PEDIGREE ANALYSIS SYMBOLS
Male
Female
Affected individuals
Mating
Parents and children (3 boys, 1 girl)
Autosomal recessive: Involves a recessive allele on a non-sex chromosome
Autosomal dominant: Involves a dominant allele on a non-sex chromosome
X-linked recessive: Involves a recessive allele on the X-chromosome
X-linked dominant: Involves a dominant allele on the X-chromosome
Y-linked: Involves an allele on the Y-chromosome

CHALLENGE: Match these disorders to an appropriate pedigree pattern above, if possible.

a. Cystic fibrosis
b. Down syndrome
c. Hemophilia
d. Lyme disease
e. Sickle cell anemia
f. Tay-Sachs disease
g. Familial adenomatous polyposis (FAP)
h. Huntington disease
i. Duchenne muscular dystrophy
j. Red-green color blindness
k. Spinocerebellar ataxia type 1 (SCA1)